APPENDIX A

Nebraska Newborn Screening and Genetics State Genetics Grant Advisory Council

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APPENDIX B

Survey of Physicians Regarding the Nebraska Newborn Screening Program

Surveys were mailed to 140 of the 840 physicians (pediatricians and family practitioners) who were listed with the NE NBS Program as having cared for infants who had NBS. Of the 140 mailed surveys, 50 were returned: a respectable response rate of about 36%. The number of responses for each answer, and the corresponding percentage for that answer, plus any comments that were given, are in bold.

In summary, only 68% of the responding physicians indicate that they have received the NBS results for an infant in their care. Other areas of NBS which the physicians have been involved include, but are not limited to: "performed/coordinated follow-up testing on a presumptive positive newborn" (54%), "learned about NBS from newsletters" (52%), "track all newborns in their care to ensure that they have been screened" (46%), and "learned about NBS from the annual report" (38%). A great majority (82%) have not attended an educational program on the NE NBS program, however, of those physicians who did attend a program did so at the local hospital, health care provider meeting at the state level, or in residency. Sixty-two percent of the responding physicians indicated that they were "somewhat comfortable" (40%) or "very comfortable" (22%) with their level of knowledge about the NE NBS program; 28% were "neutral" and 8% were "somewhat uncomfortable", while none responded that they were "very uncomfortable". Again, 62% know where to get information about the NBS program, requirements, and guidelines. A little more than half (54%) of the responding physicians indicated that they were "somewhat comfortable", and 14% were "very comfortable", with their level of knowledge about the diseases screened for by the program; 18% were "neutral", 10% were "somewhat uncomfortable", and none were "very uncomfortable". Almost three quarters of the respondents would be "very comfortable" (30%) or "somewhat comfortable" (44%) contacting parents of a newborn with a presumptive positive screen result to discuss the results and the need for follow-up testing. Twenty-eight of the physicians (56%) have had to contact parents of a newborn with a presumptive positive screen, of which, 13 (46% of the 28) provided the parents with the patient education brochure about the disorder for which the newborn screened positive. And, of those 13 physicians who provided the patient brochure, about half said the parents requested additional information regarding the initial results or disease for which the newborn screened positive. Forty-six percent of the respondents provided additional information to the parents either by giving them more information (16%) or referring them to another resource (6%) or doing both (24%). The sources where the accessed additional information included, but is not limited to: personal knowledge (36%), contacted the pediatric specialist knowledgeable about the disease (32%), medical textbooks (26%), and the Internet (10%). Half of the respondents would like the NE NBS programs to provide information on "how to explain presumptive positive screening results with sensitivity to parental anxiety while ensuring quick action to obtain confirmatory tests" and " when to refer patients to pediatric subspecialists". In addition, responding physicians requested patient education materials in Spanish, Vietnamese, Laotian, Arabic, Sudanese, and English. More than half of the respondents were from a metropolitan area (54%) and 72% were from Eastern Nebraska.

Please read the questions carefully and circle the appropriate response or responses.

1. My involvement with the newborn (metabolic) screening program has included: (Circle all the apply)

a.	Learned about the program in an in-service	**7 (14%)**
b.	Learned about newborn screening from newsletters	**26 (52%)**
c.	Learned about newborn screening from the annual report	**19 (38%)**
d.	I track all newborns in my care to ensure that they have been screened	**23 (46%)**
e.	Received results for an infant in my care	**34 (68%)**
f.	Performed/coordinated follow-up testing on a presumptive positive newborn	**27 (54%)**
g.	Provided ongoing care for a child diagnosed by the newborn screening program	**12 (24%)**
h.	Other, please specify: _____	**3 (6%)**

For "Other": "Dr. Perry", "specialists", "have had 7000 over 21 years"

****1 did not answer (2%)**

2. Have you attended an educational program on the Nebraska Newborn Screening Program?

a.	Yes	**7 (14%)**
b.	No	**41 (82%)**

**** 2 did not answer (4%)**

If yes, where was the program given? **Percentages based on 7 "yes" responses.**

i.	Local clinic	**0 (0%)**
ii.	Local hospital	**4 (57%)**
iii.	Health care provider meeting at the state level	**4 (57%)**
iv.	National conference	**0 (0%)**
v.	Other, please name: _____	**2 (29%)**

For "Other": both responses were "residency"

3. Do you feel comfortable with your level of knowledge about the Nebraska newborn screening process?

a. Very comfortable **11 (22%)**
b. Somewhat comfortable **20 (40%)**
c. Neutral **14 (28%)**
d. Somewhat uncomfortable **4 (8%)**
e. Very uncomfortable **0 (0%)**

Why or why not?

Comment: "Just finished residency"

****1 did not answer (2%)**

4. Do you know where to get information about the newborn screening program, requirements and guidelines?

a. Yes **31 (62%)**
b. No **17 (34%)**

****2 did not answer (4%)**

5. Do you feel comfortable with your level of knowledge about the diseases screened for by the Nebraska newborn screening program?

a. Very comfortable **7 (14%)**
b. Somewhat comfortable **27 (54%)**
c. Neutral **9 (18%)**
d. Somewhat uncomfortable **5 (10%)**
e. Very uncomfortable **0 (0%)**

Why or why not?

Comment: "Just moved to Nebraska"

****2 did not answer (4%)**

6. Do you think you would feel comfortable contacting parent(s) of a newborn with a presumptive positive screen result to discuss the results and the need for follow-up testing?

a. Very comfortable **15 (30%)**
b. Somewhat comfortable **22 (44%)**
c. Neutral **4 (8%)**
d. Somewhat uncomfortable **5 (10%)**
e. Very uncomfortable **0 (0%)**

Why or why not?

Comment: "Don't have enough information/knowledge"

****4 did not answer (8%)**

7. Have you had to contact parent(s) of a newborn with a presumptive positive screen?

a. Yes **28 (56%)**
b. No **22 (44%)**

8. If you answered "yes" to question 7, did you provide to the parent(s) the patient education brochure about the disorder for which the newborn had screened positive? **Percentages based on the 28 "Yes" responses in question 7.**

a. Yes **13 (46%)**
b. No **15 (54%)**

Comment: "Did not have brochure"

****22 did not answer (44%)**

9. If you answered "yes" to question 8, did the parents of the presumptive positive newborn request further information regarding the initial test results or disease for which their newborn tested positive? **Percentages based on the 13 "Yes" responses from question 8.**

a. Yes **7 (54%)**
b. No **7 (54%)**

****36 did not answer (72%)**

10. Did you provide additional information to the parents or refer the parents to another resource? (Circle one)
    a. Yes, gave the parents more information **8 (16%)**
    b. Yes, referred the parents to another resource. **3 (6%)**
    c. Yes, gave the parents more information and referred them to another resource. **12 (24%)**
    d. No **5 (10%)**

Comment: "Many parents access the internet on their own"

****22 did not answer (44%)**

11. Where did you access further information for the parents? (Circle all that apply)
    a. Personal knowledge **18 (36%)**
    b. Newborn Screening Practitioner's Manual **1 (2%)**
    c. I contacted the Newborn Screening Program **3 (6%)**
    d. I contacted the pediatric specialist knowledgeable about the disease **16 (32%)**
    e. Medical textbooks **13 (26%)**
    f. Internet **5 (10%)**
    g. Colleagues **3 (6%)**
    h. Other, please list: _____ **1 (2%) "pathologist"**

****20 did not answer (40%)**

12. What information would you like the Newborn Screening Program to provide: (Circle all that apply)
    a. How to explain presumptive positive screening results with sensitivity to parental anxiety while ensuring quick action to obtain confirmatory tests. **25 (50%)**
    b. When to refer patients to pediatric sub-specialist. **25 (50%)**
    c. Patient education materials to provide to prospective parents during the 3rd trimester to inform them of the results. **17 (34%)**
    d. Patient education materials in these languages: **23 (26%)**
        i. Spanish 17 (74%)
        ii. Vietnamese 7 (30%)
        iii. Laotian 1 (4%)
        iv. Chinese 0 (0%)
        v. Russian 0 (0%)
        vi. Arabic 1 (4%)
        vii. Other, please specify: _____ **3 (13%) (Sudanese-2, English-1)**
    e. Professional education covering the fatty acid, organic acid and amino acid disorders that can be discovered by tandem mass spectrometry (currently supplemental) screening. **13 (26%)**
    f. Professional education on the nuances of tandem mass spectrometry screening, (e.g. the effect of TPN, early gestation, prematurity, etc.) on the specimen collection time and results. **6 (12%)**

****6 did not answer (12%)**

13. Please circle the response that best describes the size of the community you live in:
    a. Metropolitan **27 (54%)**
    b. Population greater than 50,000 but outside of Omaha **1 (2%)**
    c. Population between 20,000 and 50,000 **6 (12%)**
    d. Population between 5,000 and 20,000 **8 (16%)**
    e. Population less than 5,000 **8 (16%)**

14. Please circle the response that best describes the location of the community you live in:
    a. Eastern Nebraska **36 (72%)**
    b. Central Nebraska **11 (22%)**
    c. Western Nebraska **3 (6%)**

Comments:

"No longer care for newborns" (still answered some questions)-3 (6%)

APPENDIX C

Survey of Parents of Children with Presumptive Positive Results by Newborn Screening

Surveys were mailed to 218 parents whose infants had presumptive positive results by the initial newborn screening from May to October 2001. This group was chosen because the newborn screening process was a recent event, thus the recall of the results and process would be improved, and the chance that there had been a change of address since the NBS process would be smaller. **Of the 218 mailed surveys, 20 were returned**: a disappointing response rate of about 9%. The number of responses for each answer, and the corresponding percentage for that answer, plus any comments that were given, are in bold.

In summary, three-fourths of the respondents had received information regarding newborn screening at the hospital, with 60% of those getting the information from a brochure, 53% the nurse explained, and 47% received the information orally. Again, three-fourths of the respondents understood what disorders their baby was screened for and 60% did not have questions about the NBS testing. Eighty percent of the respondents were informed of their baby's presumptive positive screening result by either their baby's pediatrician (50%) or their family doctor (30%). The information about the presumptive positive result came by a call from the doctor's office (60%), explained by the baby's doctor (50%), during scheduling for follow-up testing (25%), explained by a pediatric specialist (20%), from a brochure (10%), the internet/websites (10%), or from their work (10%). Seventy percent received the results of the follow-up testing. Fewer parents were referred to a pediatric specialist familiar with their child's condition (7 responses, 35%), than not (8 responses, 40%), although 9 respondents replied that their baby's care is followed by a pediatric specialist familiar with their child's condition. Only 5 respondents indicated that they were told about the availability of genetic services, and only 3 indicated that they did see a geneticist and/or a genetic counselor. Six respondents indicated that they would be interested in speaking with another parent who has had a similar experience, and 4 were not interested. Ninety-five percent of those responding had health insurance for their baby; 53% through an employer plan, 32% through Medicaid, 11% through SCHIP (NE Kid's Connection), and 5% through a private plan. The greatest response came from those parents living in a metropolitan area (35%) and a majority were from Eastern Nebraska (55%).

Please read the question carefully and circle the appropriate response or responses.

1. Did you receive information at the hospital about newborn screening for certain diseases?
   - a. Yes **15 (75%)**
   - b. No **4 (20%)**

   **** 1 did not answer (5%)**

   If yes, how was this information presented? (Circle all that apply) **Percentages based on 15 "Yes" responses.**
   - i. Orally **7 (47%)**
   - ii. In a brochure **9 (60%)**
   - iii. The doctor explained **6 (40%)**
   - iv. The nurse explained **8 (53%)**
   - v. The lab tech explained **1 (7%)**
   - vi. The social worker explained **0 (0%)**
   - vii. The video in the hospital explained **0 (0%)**

2. Did you understand what disorders your baby was being screened for?
   - a. Yes **15 (75%)**
   - b. No **4 (20%)**

   Comment: "After we received a positive result for biotinadase deficiency"
   ****1 did not answer (5%)**

3. Did you have questions about the newborn screening testing?
   - a. Yes **6 (30%)**
   - b. No **12 (60%)**

   **** 2 did not answer (10%)**

   If yes, were these answered to your satisfaction? **Percentages based on 6 "Yes" responses.**
   - i. Yes **4 (67%)**
   - ii. No **2 (33%)**

4. For which disorder did your child receive a presumptive positive result on the Nebraska Newborn Screen?
   a. Congenital Primary Hypothyroidism **4 (20%)**
   b. Galactosemia **2 (10%)**
   c. Hemoglobinopathies (including sickle cell anemia, thalassemia, etc…) **10 (50%)**
   d. Biotinidase Deficiency **2 (10%)**
   e. Phenylketonuria (PKU) **0 (0%)**

   ****2 did not answer (10%)**

5. Who notified you of the presumptive positive screening result? (Circle all that apply)
   a. Hospital lab or delivery staff **4 (20%)**
   b. My baby's pediatrician **10 (50%)**
   c. Our family doctor **6 (30%)**
   d. Other_____ **2 (5%) (neonatologist-1, letter at home-1)**

6. How were you informed about the presumptive positive screening result? (Circle all that apply)
   a. Received a brochure from the doctor **0 (0%)**
   b. Received a brochure from someone else **2 (10%)**
   c. My doctor's office called me **12 (60%)**
   d. During scheduling for follow-up testing **5 (25%)**
   e. Information was explained by my baby's doctor **10 (50%)**
   f. Information was explained by a pediatric specialist **4 (20%)**
   g. The internet/websites **2 (10%)**
   h. The library **0 (0%)**
   i. I got more information from: _____ **2 (10%) (at work-2)**

   Comment: "Doctor left message on home answering machine"

7. Did you receive the results of the follow-up testing?
   a. Yes **14 (70%)**
   b. No **3 (15%)**

   Comment: "It took longer than it should have and no one had any answers or knew how to read the test results."
   ****3 did not answer (15%)**

   If the results were negative, please skip questions 9-14.

8. If the results of the second test were positive, what topics were discussed with you by your baby's primary care doctor? (mark all that apply)
   a. The disorder with which your baby was diagnosed. **12 (60%)**
   b. The treatments for the disorder with which your baby was diagnosed. **6 (30%)**
   c. The future treatments and development for your baby. **8 (40%)**
   d. No education was provided. **2 (10%)**
   e. Other_____ **3 (15%)**

   Comments: "Very little information" "Everything including meds, treatment, test, etc…"
   ****8 did not answer (40%)**

9. Were you satisfied with the amount and quality of the information you received?
   a. Yes **12 (60%)**
   b. No **4 (20%)**

   ****4 did not answer (20%)**

10. Were you referred to a pediatric specialist who was familiar with your child's condition?
    a. Yes **7 (35%)**
    b. No **8 (40%)**

    Comment: "Hematologist"
    ****5 did not answer (25%)**

11. Is your baby's care being followed by a pediatric specialist familiar with your child's condition?
    a. Yes **9 (45%)**
    b. No **6 (30%)**

    ****5 did not answer (25%)**

12. Did any health care provider tell you about the availability of genetic services?
    a. Yes **5 (25%)**
    b. No **10 (50%)**

    ****5 did not answer (25%)**

13. Did you see a geneticist and/or a genetic counselor?
    a. Yes **3 (15%)**
    b. No **12 (60%)**

    Comment: "Spoke with and met with the director of the pediatric metabolism at UNMC"
    ****5 did not answer (25%)**

14. If a parent support network were available, would you like to be contacted by a representative (another parent who has had a similar experience with a child who has the same condition as your baby)?
    a. Yes **6 (30%)**
    b. No **8 (40%)**

    ****6 did not answer (30%)**

15. Do you have health insurance for your baby?
    a. Yes **18 (95%)**
    b. No **0 (0%)**

    If yes, what kind of insurance is it?
    i. Private **1 (5%)**
    ii. Employer plan **10 (53%)**
    iii. Medicaid **6 (32%)**
    iv. State Children's health Insurance Plan **2 (11%)**

    ****2 did not answer (10%)**

16. Please circle the response that best describes the size of the community you live in:
    a. Metropolitan **7 (35%)**
    b. Population greater than 50,000 but outside of Omaha **2 (10%)**
    c. Population between 20,000 and 50,000 **5 (25%)**
    d. Population between 5,000 and 20,000 **2 (10%)**
    e. Population less than 5,000 **2 (10%)**

    ****2 did not answer (10%)**

17. Please circle the response that best describes the location of the community you live in:
    a. Eastern Nebraska **11 (55%)**
    b. Central Nebraska **2 (10%)**
    c. Western Nebraska **4 (20%)**

    ****3 did not answer (15%)**

18. Do you have any suggestions to improve the delivery of parent education about Newborn Screening in Nebraska? Please explain your response.

Comments:

"Please inform parents with all pamphlets, documentation, etc… regarding any/all tests being done regardless of the results. "No news is good news" is a theory going out the door."

"I'm very thankful for this new screening, its saving a lot of lives, if only this was a givin in every state to do the screening upon birth. Hats off to the doctor in the genetics hospital in Omaha. Follow up on positive results and further information on the disease would be helpful. Not much was discussed since no one in the office understood the information from Omaha-Doctor just said wait for more information and don't worry."

"Internet very helpful"

"I was told before further testing that I should stop pumping because the test came back positive, then a month later further results showed negative and I could no longer breast feed. The doctor really knew nothing about this."

"We wish there was a specialist closer to our area, as no doctors treat for primary hypothyroidism. Until we saw the specialist we felt alone, not really knowing that we were doing everything we could. Thanks to this screening our child is perfectly normal. Had this screening been an option we may not have done it!"

*****Two surveys came back with several questions not having been printed on them*****

APPENDIX D

Survey of Parents Regarding the Nebraska Newborn Screening Program

Surveys were mailed to 268 parents whose infants had newborn screening from May to October 2001. This group was chosen because the newborn screening process was a recent event, thus the recall of the results and process would be improved, and the chance that there had been a change of address since the NBS process would be smaller. **Of the 268 mailed surveys, 38 were returned**: a response rate of about 14%. The number of responses for each answer, and the corresponding percentage for that answer, plus any comments that were given, are in bold.

In summary, 45% of the respondents indicated that they had never learned about the NE NBS program and another 21% first learned about the program from the newborn packet received at the hospital. Only 4 respondents (11%) first learned of the NBS program during a visit with their obstetrician or family doctor. Thirty-two percent learned what disorders the screening can detect and 29% learned how the blood sample is obtained. Those parents who did learn about the NBS received their information from the hospital staff (18%) and printed brochure (18%), as well as through their family doctor (16%) or another doctor (8%). Eleven respondents (29%) indicated a "fair" response to the question asking if the materials presented by the hospital staff and physician were understandable (53% indicated that this question was "not applicable"). In response to the question about whether the brochure was understandable, 61% indicated "not applicable", 7 marked "good", 6 "fair", and 2 "very good". Equal numbers of respondents (12 each) who had questions after reading the brochure indicated that they were and weren't able to get answers to those questions. Eight-three percent of those who were able to get answers to their questions received them from their baby's doctor; one respondent also received answers from the State NBS program. Although 39% of the respondents indicated that they did not understand the reasons why NBS is done for every baby in NE (58% did understand), 76% agreed that NBS should be mandatory (16% did not). Sixty-three percent of respondents were not given the results of their baby's NBS. Nearly half (47%) of the responses were from parents living in communities with a population less than 5,000 (21% were metropolitan, 18% from communities with a population between 5,000 and 20,000), and half were returned from Eastern NE (29% from Central NE, 18% from Western NE, 3% from Iowa).

Please read the questions carefully and circle the appropriate response or responses.

1. When did you first learn about the Nebraska Newborn Screening Program? (Choose one)
   - a. During a second trimester visit with my obstetrician or family doctor **1 (3%)**
   - b. During a third trimester visit with my obstetrician or family doctor **3 (8%)**
   - c. When admitted to the hospital for delivery **2 (5%)**
   - d. In the newborn packet received at the hospital **8 (21%)**
   - e. In the newborn packet read at home **2 (5%)**
   - f. Upon leaving the hospital **1 (3%)**
   - g. Never **17 (45%)**
   - h. Other, please specify: _____ **4 (11%)**

   For "Other": "don't know what it is," "during birth class," "when I moved to town," "after my child was born"

2. What did you learn about the process of Newborn Screening? (Circle all that apply)
   - a. How the blood sample is obtained **11 (29%)**
   - b. What disorders the screening can detect **12 (32%)**
   - c. What "metabolic" and "genetic" disorders mean **6 (16%)**
   - d. How you will be notified of a presumptive positive screen result **7 (18%)**
   - e. Who will notify you of a presumptive positive screen result **6 (16%)**
   - f. Did not learn about the process of Newborn Screening **19 (50%)**
   - g. Other, please specify: _____ **3 (8%)**

   For "Other": "not sure unless it was part of the screening done at the hospital," "didn't read," "it was difficult to obtain the information regarding the supplemental screening"
   ****1 did not answer (3%)**

3. How was the information about the Newborn Screening Program presented? (Circle all that apply)
   a. Through your family doctor **6 (16%)**
   b. Through another doctor, please specify that doctor's specialty: _____ **3 (8%)**
   c. Through hospital staff **7 (18%)**
   d. Through printed brochure **7 (18%)**
   e. Was not presented information **17 (45%)**
   f. Other, please specify: _____ **3 (8%)**

   For "Other": "didn't read," "my Ob/Gyn got the information after I requested it," and no answer written
   Comment: "Maybe the hospital did give us the information, but it wasn't pointed out or discussed with us"

4. How understandable were the materials presented by hospital staff and physician?
   a. Very good **2 (5%)**
   b. Good **5 (13%)**
   c. Fair **11 (29%)**
   d. Poor **0 (0%)**
   e. Very poor **0 (0%)**
   f. Not applicable **20 (53%)**

   Comments: "Nothing was mentioned at the hospital, just got a list of what was screened for" and "I'm Spanish"

5. How understandable was the information in the printed brochure?
   a. Very good **2 (5%)**
   b. Good **7 (18%)**
   c. Fair **6 (16%)**
   d. Poor **0 (0%)**
   e. Very poor **0 (0%)**
   f. Not applicable **23 (61%)**

   Comment: "I read Spanish"

6. If you had questions after reading the brochure, were you able to get answers?
   a. Yes **12 (32%)**
   b. No **12 (32%)**

   If yes, who answered your questions? **Percentages based on 12 "yes" responses.**
      i. My baby's doctor **10 (83%)**
      ii. The State Newborn Screening Program **1 (8%)**
      iii. Other, please specify: _____ **1 (8%) (no brochure-1)**

   ****13 did not answer (34%)**

7. Do you understand the reasons why Newborn Screening is done for every baby born in Nebraska?
   a. Yes **22 (58%)**
   b. No **15 (39%)**

   ****1 did not answer (3%)**

8. Do you agree that Newborn Screening for metabolic disorders should be mandatory in Nebraska?
   a. Yes **29 (76%)**
   b. No **6 (16%)**

   ****3 did not answer (8%)**

9. Were you given the results of your baby's Newborn Screening?
   a. Yes **12 (32%)**
   b. No **24 (63%)**

Comments: "It was sent to the doctor," "I was told I would get results in the mail and I never did-I had to ask at her next doctor's appointment," "Never saw the results, but was told verbally"

****2 did not answer (5%)**

10. Please circle the response that best describes the size of the community you live in:
    19. Metropolitan **8 (21%)**
    20. Population greater than 50,000 but outside of Omaha **2 (5%)**
    21. Population between 20,000 and 50,000 **3 (8%)**
    22. Population between 5,000 and 20,000 **7 (18%)**
    23. Population less than 5,000 **18 (47%)**

11. Please circle the response that best describes the location of the community you live in:
    24. Eastern Nebraska **19 (50%)**
    25. Central Nebraska **11 (29%)**
    26. Western Nebraska **7 (18%)**

    ****Iowa not given as a choice, but 1 response from Iowa (3%)**

APPENDIX E

State Genetics Program Self-Assessment Tool

(Modified from the PacNoRGG Assessment Tool with reference to NNSGRC Guidance (See Community Genet 2001;4:175-196)

I. ORGANIZATION & ADMINISTRATION			In Place	Comments (i.e. planned, etc.)
A. Genetics is recognized as a separate program			☐	**In services, not in public health**
B. A full-time genetics coordinator is employed or provided for			☐	**No**
C. A needs assessment has been conducted			☐	**Yes**
D. A formal plan for integrating genetic services into public health exists			☐	**No, some plans forthcoming**
E. The plan organized by public health core functions?			☐	**No, current strategy is just to look at infrastructure needs**
1.	**Assessment -** *Does the plan describe:*		☐	**Needs assessment is in progress (IP)**
	a.	The state (size, geography, industry, etc.)	☐	**(IP)**
	b.	Demographic parameters (age, race, socioeconomic status, etc.)	☐	**(IP)**
	c.	Public health & genetics-related systems and needs	☐	**(IP)**
	d.	Data collection system	☐	**(IP)**
	e.	System for evaluation of genetics services	☐	**(IP)**
	f.	System for evaluation of educational activities	☐	**(IP)**
	g.	*Other:*	☐	**(IP)**
2.	**Policy Development -** *Does the plan describe:*		☐	**(IP)**
	a.	Legislation or Rules in place related to genetics	☐	**(IP)**
	b.	Mechanisms of funding/reimbursement for genetic services	☐	**(IP)**
	c.	*Other:*	☐	**(IP)**
3.	**Assurance -** *Does the plan include strategies to assure:*		☐	**(IP)**
	a.	A network of genetic services	☐	**(IP)**
	b.	A system of prevention services	☐	**(IP)**
	c.	Genetics education activities	☐	**(IP)**
	d.	Periodic review of genetic services	☐	**(IP)**
	e.	A framework for existing quality assurance measures for clinical and laboratory services	☐	**(IP)**
	f.	*Other:*	☐	

4.	**Collaboration –** *Is there collaboration with relevant groups:*		☐	**Yes**
	a.	*Consumers*	☐	**Yes**
	b.	*Researchers*	☐	**Yes**
	c.	*Genetics Services Providers*	☐	**Yes**
	d.	*Teachers*	☐	**No**
	e.	*Other:*	☐	**Legislators**
F. There is meaningful input from consumers and underserved populations in program planning			☐	**In needs assessment stage, and somewhat ongoing w/ tech. Committee**
G. There are links to other health programs with genetics components			☐	
	1.	*Chronic Disease*	☐	**No**
	2.	*Epidemiology*	☐	**No**
	3.	*Environmental Health*	☐	**No**
	4.	*Other:*	☐	
	5.	*Other:*	☐	

II. SERVICES

A.	**Family Based Services**		***Centrally Coordinated***	***Available Regionally (in state)***	***Available in Select Commun.***	***Coord./ Availa. from Other States***	***Not Coord. with Other States/ Not Avail.***	***Comments***
	1.	General Genetic Clinics	☐	☐	**x**	☐	☐	
	2.	Metabolic Clinics	**x**	☐	☐	☐	☐	
	3.	Single Disease Clinics	☐	☐	☐	☐	☐	
		a. Hemophilia	**x**	☐	☐	☐	☐	
		b. Cystic Fibrosis	**x**	☐	☐	☐	☐	
		c. Other:	☐	☐	☐	☐	☐	
	4.	Prenatal Genetics Clinics	**x**	☐	☐	☐	☐	

B.	Population Based Services		Centrally Coordinated	Available Regionally (in state)	Available in Select Commun.	Coord./ Available in Other States	Not Coordinated with Other States/Not Available	Comments
	1.	Preconception screening	☐	☐	☐	☐	☐	
	2.	Prenatal Screening	x	☐	☐	☐	☐	
		a. *Maternal*	☐	☐	☐	☐	☐	
		b. *AMA*	☐	☐	☐	☐	☐	
		c. *Family History*	☐	☐	☐	☐	☐	
		d. *Carrier screen for targeted populations*	☐	☐	☐	☐	☐	
		e. *Other:*	☐	☐	☐	☐	☐	
	3.	Newborn Screening	☐	☐	☐	☐	☐	
		a. *Lab Services*	☐	x	☐	☐	☐	
		b. *Follow Up*	x	☐	☐	☐	☐	
	4.	Childhood	☐	☐	x	☐	☐	
	5.	Adult Screening	☐	☐	x	☐	☐	

C.	Clinical Laboratory Services		Available In-State	Coordinated with Out-of-State Lab	Not Available/Not Coordinated	Comments
	1.	Cytogenetics	x	☐	☐	
	2.	Biochemical Genetics	x	☐	☐	
	3.	Molecular Genetics	x	☐	☐	

D.	Modes of Service Delivery		Check all that apply
	1.	Genetics Unit of State Health Department	☐
	2.	Large, comprehensive genetics center	X
	3.	Genetics unit of a comprehensive managed care facility	☐
	4.	Resident genetics unit within a primary health care facility	☐
	5.	Resident board certified genetic counselor and/or PhD medical geneticist with periodic visits by a board certified MD Medical Geneticist	☐
	6.	Periodic visits by a board certified genetic counselor, medical geneticist, or other staff with local coordinators at outreach clinics	X
	7.	Genetics clinics in the private sector conducted by trained MD geneticists	X
	8.	Board certified genetic counselor and/or PhD medical geneticist within single disease/medical specialty setting	☐
	9.	Other:	☐

E.	Genetics Professionals		Centrally Coordinated	Available Regionally	Available in Select Commun.	Coord./ Available in Other States	Not Coordinated/ Not Available	Comments
	1.	Clinical Geneticist	x	☐	☐	☐	☐	
	2.	PhD Medical Geneticist	x	☐	☐	☐	☐	
	3.	Genetic Counselor	x	☐	☐	☐	☐	
	4.	Clinical Cytogeneticist	x	☐	☐	☐	☐	
	5.	Clinical Biochemical Geneticist	x	☐	☐	☐	☐	
	6.	Clinical Molecular Geneticist	☐	☐	☐	☐	x	
	7.	Cytogenetic Technologist	x	☐	☐	☐	☐	
	8.	Genetics Nurse	x	☐	☐	☐	☐	
	9.	Advance Practice Nurse in Genetics	x	☐	☐	☐	☐	
	10.	Perinatologist/ Obstetrician	☐	☐	x	☐	☐	
	11.	Dietician	x	☐	☐	☐	☐	
	12.	Other:	☐	☐	☐	☐	☐	

III. FUNDING OF SERVICES		Check all that apply
A.	Medicaid, Medicare reimbursement	X
B.	Third Party Carriers reimbursement	X
C.	Newborn Screening Surcharge	X
D.	State General Funds	X
E.	Federal Title V (MCH Block Grant)	X
F.	Other State and/or Federal Grants	X
G.	Specific Disease-Related Organizations	☐
I.	Other:	☐

IV. DOCUMENTATION OF NEEDS & SERVICES						
A.	**Data Sources**		***Available; Utilized***	***Available; Not Utilized***	***Not Available***	***Comments***
	1.	State level clinical genetics database	☐	☐	x	
	2.	Newborn screening database	x	☐	☐	
	3.	Vital statistics: birth, fetal death, death certificates	x	☐	☐	
	4.	Statewide hospital discharge data	x	☐	☐	
	5.	Medicaid/Medicare eligibility, claims, provider datasets	☐	x	☐	
	6.	Local/Statewide/Regional cytogenetics registry	☐	x	☐	
	7.	Local/Statewide/Regional birth defects registry	☐	x	☐	
	8.	Local/Statewide/Regional population based cancer/tumor registry	☐	x	☐	
	9.	Directory of genetic service providers and referral sources	x	☐	☐	
	10.	Cytogenetics laboratory databases collected by ACT	x	☐	☐	
	11.	Federal census data	x	☐	☐	
	12.	Special surveys and projects:	x	☐	☐	
		a. Pregnancy Risk Assessment Monitoring System (PRAMS)	x	☐	☐	
		b. National Maternal & Infant Health Survey	☐	☐	x	
		c. Behavioral Risk Factor Surveillance System (BRFSS)	☐	x	☐	
		d. National Survey of Family Growth	☐	☐	x	
	13.	Other:	☐	☐	☐	

B.	Data Linkages		*Linkage in Place*	*Planned Linkage*	*Linkages Being Considered*	*Comments*
	1.	Birth & death for all deaths up to six years of age	☐	x	☐	
	2.	Birth defects & tumor registry for all pediatric cancer cases	☐	☐	x	
	3.	Birth defects registry records with vital statistics	x	☐	☐	
	4.	Inpatient hospital discharge records with birth certificates	x	☐	☐	
	5.	Newborn screening records with birth certificates	x	☐	☐	
	6.	MSAFP/AFAFP/Triple Screen with vital statistics	☐	☐	☐	No
	7.	Statewide clinical genetics services database and birth/fetal death certificates (in form of numerator/denominator ratio)	☐	☐	x	
	8.	System for direct referral from clinical genetics to early intervention services for infants < 3 years of age; Children with Special Health Care Needs (CSHCN); Supplemental Social Insurance (SSI); etc.	x	☐	☐	
	9.	Other:	☐	☐	☐	

V. PREVENTION

A.		Primary Prevention Programs	*In Place*	*Planned*	*Comments*
	1.	*Folic Acid Education*	x	x	
	2.	*Teratogen Information Services*	x	☐	
	3.	*Other:*	☐	☐	
	4.	*Other:*	☐	☐	
	5.	*Other:*	☐	☐	
	6.	*Other:*	☐	☐	
	7.	*Other:*	☐	☐	
	8.	*Other:*	☐	☐	

B.		Secondary Prevention Programs	*Readily available to all state residents*	*Readily available to most state residents*	*Readily available to some state residents*	*Not readily available to state residents*	*Comments*
	1.	Preconception Screening	☐	☐	☐	☐	
	2.	Prenatal Screening	☐	x	☐	☐	
	3.	Newborn Screening	x	☐	☐	☐	
	4.	Childhood Screening	☐	☐	x	☐	
	5.	Adult Screening	☐	☐	x	☐	

C.		Tertiary Prevention Programs	*Check all that apply*
	1.	*Educational and other special services for individuals with special needs*	X
	2.	*Appropriate management of genetic disorders*	X
	3.	*Access to medical devices*	X
	4.	*Referral of families to support groups; or facilitation of contact with similarly affected families*	X
	5.	*Other:*	☐
	6.	*Other:*	☐
	7.	*Other:*	☐
	8.	*Other:*	☐
	9.	*Other:*	☐

MODIFICATION EXAMPLES

#1 your state may have examples to be added in some sections. Here, Section IIIB has been modified:

B.	Population Based Services					
	1.	Prenatal Screening	☐	☐	☐	☐
		Maternal Serum Screening	☐	☐	☐	☐
		Maternal Age	☐	☐	☐	☐
	2.	Newborn Screening	**x**	☐	☐	☐
		CH	**x**	☐	☐	☐
		PKU	**x**	☐	☐	☐
		Hemoglobinopathies	**x**	☐	☐	☐
		CAH	☐	☐	☐	☐
	3.	Childhood Screening	☐	☐	☐	☐
	4.	Adult Screening	☐	☐	☐	☐

#2 You may also wish to modify the tool in other ways. For example, you may want to change assessment parameters in some sections. Here, Section IIIE has been modified:

E.	Genetics Professionals		*FTEs - Residing in Community Based Clinics*	*FTEs Traveling to Outreach Clinics*	*FTEs In Comprehensive Centralized Genetics Centers*	*Ratio: FTE/population*
	1.	Clinical Geneticist	☐	☐	☐	7
	2.	PhD Medical Geneticist	☐	☐	☐	
	3.	Genetic Counselor	☐	☐	☐	8
	4.	Clinical Cytogeneticist	☐	☐	☐	
	5.	Clinical Biochemical Geneticist	☐	☐	☐	4
	6.	Clinical Molecular Geneticist	☐	☐	☐	yes
	7.	**Total State Population** (this number can be used to obtain ratio of state residents: genetic professional)				**1.7 million**

APPENDIX F

Collaboration Survey Results from June 2002 as compared to the results of September 2001

The following survey was again given to the members of the Nebraska Newborn Screening and Genetics State Advisory Committee (NBSGAC) in June 2002, ten months after the original distribution. In September 2001, 45 members of the NBSGAC were surveyed regarding their feelings about how children with special health care needs (CSHCN) were being served in Nebraska. The committee consists of physicians, pediatric specialists, parent consumers, and employees of various departments of the Nebraska Health and Human Services System. Thirteen of the 45 anonymous surveys were returned resulting in a 29% response rate. In June 2002, 43 surveys were mailed to the committee members, and 23 were anonymously returned for a 53% response rate.

The **bold** numbers are the responses from June 2002, and the *italicized* numbers are from September 2001. The percentage totals, given at the end of the statements, do not always equal 100% due to rounding. The results are as follows:

1. It appears/seems to me that *public health agencies* and *providers* collaborate well in providing/assuring services to CSHCN. **(100%)** *(100%)*	Strongly Agree **0%** *0%*	Agree **26%** *54%*	Neutral **39%** *23%*	Disagree **35%** *23%*	Strongly Disagree **0%** *0%*	Not Applicable **0%** *0%*
2. It appears/seems to me *providers* and *consumers* collaborate well in providing services to CSHCN. **(100%)** *(100%)*	Strongly Agree **0%** *0%*	Agree **57%** *38%*	Neutral **22%** *54%*	Disagree **17%** *8%*	Strongly Disagree **4%** *0%*	Not Applicable **0%** *0%*
3. It appears/seems to me that *public health agencies* and *consumers* collaborate well in providing/assuring services to CSHCN. **(100%)** *(100%)*	Strongly Agree **0%** *0%*	Agree **39%** *23%*	Neutral **22%** *54%*	Disagree **30%** *15%*	Strongly Disagree **9%** *8%*	Not Applicable **0%** *0%*
4. I think that it is easy to locate the necessary resources for a CSHCN. **(101%)** *(100%)*	Strongly Agree **0%** *0%*	Agree **9%** *23%*	Neutral **35%** *38%*	Disagree **48%** *31%*	Strongly Disagree **9%** *0%*	Not Applicable **0%** *8%*
5. I think that it is easy to contact the necessary resources for a CSHCN. **(100%)** *(101%)*	Strongly Agree **0%** *0%*	Agree **17%** *31%*	Neutral **35%** *31%*	Disagree **48%** *31%*	Strongly Disagree **0%** *0%*	Not Applicable **0%** *8%*
6. I think that it is easy to communicate with the necessary resources for a CSHCN. **(99%)** *(101%)*	Strongly Agree **0%** *8%*	Agree **30%** *38%*	Neutral **30%** *31%*	Disagree **22%** *8%*	Strongly Disagree **4%** *8%*	Not Applicable **13%** *8%*
7. I think that resources which have been contacted on behalf of a CSHCN are prompt in providing follow-up information or care. **(100%)** *(100%)*	Strongly Agree **0%** *0%*	Agree **39%** *31%*	Neutral **39%** *46%*	Disagree **13%** *8%*	Strongly Disagree **0%** *0%*	Not Applicable **9%** *15%*
8. I have been able to obtain the necessary resources to meet the needs of a CSHCN. **(99%)** *(99%)*	Always **4%** *0%*	Usually **35%** *38%*	Sometimes **30%** *23%*	Rarely **0%** *0%*	Never **0%** *0%*	Not Applicable **30%** *38%*

Comments:
From the responses during June 2002:

1) Biggest concern: Physicians not knowing what services are available
2) I have no direct contact with a CSHCN's
3) Too bad you didn't include the 3rd party payors (BC/BS, Mutual of Omaha, etc.). They would not have scored well. I assumed that Nebraska Medicaid was included in above survey.
4) As a non-provider/non-user, I can only surmise for 1-5, and I have not attempted 6-8.
5) My perception is that public health agencies tend to ignore the needs of CSHCN and adults with special needs. Their programs and services are geared to the "typical" population.

From the responses during September 2001:

1) Need better support groups
2) Some areas collaborate, some don't
3) Disagree providers collaborate

In summary, the majority of responses continue to fall within the "agree", "neutral", and "disagree" categories with very few responses in the "strongly agree" or "strongly disagree" areas. This would again indicate that the overall feeling about how children with special health care needs are being served is fairly satisfactory or only mildly disagreeable. There has been some shift of responses from "agree" to "neutral", "disagree", and/or "strongly disagree" for the first, fourth, fifth, and sixth questions; however, there was an increase in "agree" responses for the second and third questions. The responses to the seventh and eighth questions remained roughly the same except that someone responded that they "always" have been able to obtain the necessary resources to meet the needs of a CSHCN. Once again, it appears that every area of collaboration could stand for improvement, as there has been a slight decrease of positive responses to half of the questions.

Glossary:

Collaboration – to work jointly with others or together especially in an intellectual endeavor
Public Heath Agency – provides services which are population-based which focus on improving the health status of the entire population as opposed to the treatment of individuals
Provider – trained medical professional (MD, RN, PA, midwife, genetic counselor, etc) and other allied service provider (respite care, social service, service coordination, transportation, etc)
Consumer – any child with special health care needs and their parents or guardians
CSHCN – Children with Special Health Care Needs

APPENDIX G

Guiding Principles on Human Genetic Technologies from the Report of the Nebraska Commission on Human Genetic Technologies December 1998

These Guiding Principles are designed to serve as the governing context and standard of reference for all present and future policies, practices, laws, regulations and educational initiatives related to human genetic technology in the State of Nebraska. The Nebraska Human Genetic Technologies Commission's central purpose is to encourage uses of human genetic technologies that contribute to the improvement of the human condition while assuring the protection of fundamental human rights. In these principles the term "respect" acknowledges that compelling social interests may at times require abridgement of individual liberties regarding the use of genetic technologies in order to protect the well-being of society as a whole.

Respect for Humanity

1. The inherent dignity and intrinsic value of human beings must govern all uses of human genetic technologies.
2. Human uniqueness and diversity must be respected as a cherished part of our shared humanity.

Respect for the Individual

3. Genetic information should not be used to deny individual opportunity.
4. Confidentiality and privacy concerning genetic information should be respected.
5. Individuals should be fully informed and give their voluntary consent prior to genetic testing or genetic intervention.

Respect for the Community

6. The social values of justice, equity, beneficence, do no harm, and veracity must be respected in the development and implementation of human genetic technology.
7. No group should become the subject of unfair discriminatory policies or practices on the basis of its genetic makeup.
8. Thoughtful on-going civic discourse about the role of human genetic technologies in furthering the common purposes and goals of our shared humanity is essential.

APPENDIX H

Report of the Congenital Anomalies Subcommittee of the Governor's Blue Ribbon Panel on Infant Mortality

Committee Members:
Dr. Brad Schaefer, Chair
Dr. David Bolam
Karen Heusel
Senator Jim Jensen
Roger Massey
John Wiley

Consultants to The Committee:
Dr. William Sappenfield
Dr. David Schor
Debbi Barner-Josiah

Section I Epidemiology of Congenital Anomalies (General)

A: United States Epidemiologic Data

- 3-4% of all live births have a congenital anomaly recognizable in the newborn period.
- An additional 3-4% of individuals have a congenital anomaly that is not recognizable in the neonatal period.
- Only 20% of congenital anomalies have a known etiology.
- Nationwide, the overall congenital anomaly rate is staying fairly consistent. There are, however, notable exceptions. Nationwide, congenital heart disease (most prominently atrial septal defects and hypoplastic left heart), obstructive uropathies, and neurodevelopmental disorders have been increasing.
- There is a direct correlation with the states which have a "better" tracking system and a higher-reported incidence of birth defects (correlation with better reporting).

B: Nebraska Epidemiologic Data

1. 1984-1993 Data
   - Infants with one or more birth defect noted = 4.4%
   - Rate of birth defects in 1984 (22/1000) as compared to 1993 (43.6/1000).
   - Increased rate in blacks (at least for 1990 data).
   - For this 10-year period, 7,688 infants noted with recognizable birth defects.
   - Significant increase in congenital heart disease during this 10-year period, 2063 of the 7,688 with congenital heart disease (27%)
   - No change in the rate of anencephaly.
   - Declining rate of spina bifida without anencephaly during this 10-year period (pre-folate intervention).

2. 1990-1997 Data
   - Eight year mean = 37.9/1000.
   - Birth defects rates declining at a rate of approximately 2.5% per year.
   - As seen in the national data, Nebraska's data also reflects several notable trends including an increase in specific types of congenital anomalies, despite the overall declining rate.

Notable trends includes:

1. Increase in trisomies
2. Increases congenital heart disease
3. Stable rate of anencephaly
4. Declining rate of spina bifida

- At least one collaborative study between UNMC and NDHHS documents an association of Atrazine with limb reduction defects.

Section II Congenital Anomalies and Infant Mortality

A: United States Data

- The leading cause of infant mortality in the United States:
  - For African/Americans, it represents the second most common cause, with pre-term labor and low birth weight infants being most common cause of infant mortality.
  - Also second leading cause in Native Americans and Alaskan natives.
- Infants with major congenital anomalies represent a six-fold increase incidence rate of infant deaths as compared to those without congenital anomalies.
- 45% of all NICU deaths are due to congenital anomalies with the most prevalent being:
  1. Congenital heart disease
  2. Pulmonary hypoplasia (associated with other anomalies such as diaphragmatic hernia and obstructive uropathies)
  3. "Lethal" genetic disorders — eg, trisomies and anencephaly
- Although infant mortality nationwide is decreasing, the rate at which it is decreasing is not nearly as much.
- Congenital anomalies as leading cause of infant mortality:
  1. Cardiovascular defects represent the #1 cause, representing ½ of all infant deaths due to congenital anomalies.
  2. Nationwide, a trend of infant deaths due to trisomies is significantly increased (particularly trisomies 13 and 18).
  3. Reduction of brain anomalies.
- Consistently higher birth defects rates being reported in the South and parts of the Midwest (including Nebraska) as compared to other parts of the country.

B: Nebraska Data

- Number 1 or #2 cause of infant mortality (1993): 1.8/1000 births had infant mortality due to congenital anomalies.
- 26% of infant deaths due to congenital anomalies (compared to 22% for the United States).
- The ratio of birth defects deaths to all births was 0.2% (0.6% for the United States).
- Increased neonatal deaths in Nebraska as compared to the United States for:
  1. Trisomies (2 X)
  2. Central nervous system malformations (1.6 X)
  3. Diaphragmatic hernia (1.5 X)
  4. Renal (1.89 X)
- On inspection of the data, there looks like there are specific peaks in the years 1985 and 1995.
- The overall trend is for an increasing neonatal mortality due to birth defects in Nebraska of 2.2% declining, with 3.0% for the United States.

Section III Special Issues

A: Pew Commission Recommendations

- Strengthen efforts to prevent birth defects via a national standardized approach to monitoring birth defects.
- Nebraska received a "B" grade.
- Only 10 states have "active methods" (those that received an "A" grade).
- Not enough information on environmental toxins to make definitive conclusions.

B: Infant Mortality in general does show ethnic-specific differences.

- For whites, the rate is increasing 1% per year (rate of 6.8).
- For Native Americans, it is dropping 7% per year (rate 13.1).
- For Hispanics, dropping 2% per year (rate 9.6).
- For blacks, declining 2% per year (rate 20.0).

(It has not been determined whether this is due to genetic or socio/economic factors, or both.)

Section IV Conclusions

1. The overall rate of congenital anomalies is stable. Some are decreasing (neural tube defects) and others increasing (congenital heart disease, trisomies) with the overall rate being fairly constant at around 4%.
2. Congenital anomalies are the leading cause of infant mortality nationwide and in Nebraska in whites and Hispanics, they are the second leading cause for blacks, Native Americans and Alaskan Natives.
3. Nebraska has experienced sharp increases in the infant mortality rate due to birth defects in 1986 and 1995 that make it difficult to describe recent trends. The trend in Nebraska is flat at best, and may be increasing.
    - Different linear regressions using different time periods give substantially different results.
    - Rates assuming 1,3,5,7 year averages blunt the peaks but still suggest trends
    - Using 5% and 95% confidence intervals the two peaks border on statistical significance.
4. Nebraska has a significant increase in certain types of congenital anomalies as compared to the US and a concomitant increase in infant mortality due to these anomalies.

Section V Recommendations

Preliminary thoughts: In order to effect the timely implementation of the recommendations below, it is suggested that an active, prospective study of costs and specific implementation plans be initiated for each of the noted items.

A: Short-term Recommendations

- Improve birth defects surveillance, assessment, and prevention in Nebraska.
    1. Increase funding for the Birth Defects Registry to promote an active system for assessment and educational component for contributing sites.
    2. Direct fiscal support of epidemiologic studies of:
        1. Trisomies (priority)
        2. Congenital heart disease (priority)
        3. Obstructive uropathies
- Resources should be directed toward known factors that can be modified to reduce the rate of birth defects. These would include:
    1. Educational campaign for alcohol, smoking, teratogen exposure, and drugs and medications.
    2. Folic acid awareness.

3. Maternal nutrition and health.

    - Priority areas would include diabetes mellitus, hypertension, and obesity.
    - Major focus here needs to be on pre-conceptional health.

4. Advancements should be made in the studies on environmental toxins (especially agrarian pollutants) as contributors to the occurrence of birth defects. There should be a priority given to these studies in statewide health funding, including funds such as the Excellence in Health Care Fund, etc.
5. Implementation of advanced newborn screening for disorders such as MCAD and other advanced technology (tandem spectroscopy) screening in the state of Nebraska.

B: Long-term Recommendations

- Establish a Child Health Institute with a global collaborative approach to congenital anomalies. This Institute should focus on:
    1. Policy
    2. Epidemiology
    3. Basic science and molecular biology research
    4. Environmental biology
    5. Community outreach and service
    6. Training of current and future healthcare practitioners
    7. Prevention strategies

    Such a collaborative should link together the best expertise in the state and at a minimum should include a cooperative effort of University of Nebraska Medical Center, University of Nebraska-Lincoln, Creighton University, Children's Hospital, and Health and Human Services Child Health Programs.

Respectfully submitted,

G. Bradley Schaefer, M.D., F.A.A.P., F.A.C.M.G.
for the Congenital Anomalies Subcommittee
The Governor's Blue Ribbon Panel
March 22, 2000

APPENDIX I

Healthy People 2010 Goals and Action Steps

In January 2000, the Department of Health and Human Services unveiled *Healthy People 2010.* The publication created a new model for serving children with special health care needs: community-based, family-centered, culturally competent, and coordinated care, delivered within comprehensive and integrated systems of services. More specifically, the document outlines six goals and corresponding action steps.

1. Families of children with special health care needs will partner in decision making at all levels and will be satisfied with the services they receive.
    - 1.1 Ensure that partnerships between families and professionals are a key element of the medical home.
    - 1.2 Support family-professional collaboration on efforts to improve systems of care for children and youth with special health care needs.
    - 1.3 Support the participation of traditionally underrepresented families in all decision-making, learning, and advocacy activities.
    - 1.4 Develop and promote measures for determining family satisfaction with health care services.
2. All children with special health care needs will receive coordinated, ongoing, comprehensive care within a medical home.
    - 2.1 Standardize the core elements of the medical home.
    - 2.2 Promote the medical home approach.
    - 2.3 Achieve universal access to medical homes.
    - 2.4 Use the medical home as a measure of quality care.
3. All families of children with special health care needs will have adequate private and/or public insurance to pay for the services they need.
    - 3.1 Expand insurance to all uninsured children and youth with special health care needs.
    - 3.2 Assure comprehensive coverage for children and youth with private and/or public insurance and address the issue of underinsurance.
    - 3.3 Strengthen the financing system.
4. All children will be screened early and continuously for special health care needs.
    - 4.1 Improve access to and availability of screening services.
    - 4.2 Support data capacity for integration of screening results.
    - 4.3 Improve screening guidelines and standards.
    - 4.4 Promote awareness of the need for and benefits of early and continuous screening.
5. Community-based service systems will be organized so families can use them easily.
    - 5.1 Assist communities to develop services systems that are fully inclusive of children and youth with special health care needs and their families.
    - 5.2 Build the capacities of key community stakeholders to develop community-based systems of services.
    - 5.3 Provide adequate public financing for community-based systems of services.
    - 5.4 Assure that every community has adequate services for all children with special health care needs that are organized so families can use them easily.
    - 5.5 Assure results-based accountability of community-based systems of services.
6. All youth with special health care needs will receive the services necessary to make transitions to all aspects of adult life, including adult health care, work, and independence.
    - 6.1 Use Title V to facilitate the development of Healthy and Ready to Work (HRTW)/Transition systems for children, youth, and young adults with special health care needs and their families.

6.2 Assure that youth with special health care needs participate as decision makers and as partners.
6.3 Make sure that all youth with special health care needs have accessible and affordable health insurance coverage.
6.4 Assure that all youth with special health care needs have medical homes responsive to their needs. (All Aboard the 2010 Express: A 10-Year Action Plan)

APPENDIX J

Legislative Bill 119

Statement of Intent

LB119 contains technical clarification and definitions to provisions of LB432, signed into law in 2001, an act relating to medical tests, consent to genetic testing and metabolic disease testing and the use and disposal of specimens.

Committee Statement

Summary of purpose and/or changes: The bill makes the following changes in statutes related to genetic testing and metabolic disease testing of infants:

Genetic testing. In section 71-1,104.01, the bill deletes all references to "presymptomatic genetic test" and redefines "predictive genetic test." Predictive genetic test, as redefined, means "a genetic test for an otherwise undetectable genotype or karyotype relating to the risk for developing a genetically related disease or disability, the results of which can be used to substitute a patient's prior risk based on population data or family history with a risk based on genotype or karyotype." A predictive genetic test does not include diagnostic testing diagnostic testing of a person with clinical signs or symptoms of a possible genetic condition or prenatal testing unless conducted for an adult-onset condition that is not expected to cause clinical signs or symptoms before the age of majority.

The bill removes all occurrences of the phrase "legally authorized" before the term "representative," as it refers to a person who is authorized to consent to the genetic testing of a patient on his or her behalf. The bill also adds the phrase "lacking decisional capacity" when referring to such patient for whom the consent of a representative would be permitted.

Metabolic disease testing. The bill replaces subsection (4) of section 71-519 relating to use of blood specimens collected by the Department of Health and Human Services Regulation and Licensure under such section. The bill provides that all such specimens are property of the State of Nebraska. The department is required to develop procedures for the retention, use, and disposal of such specimens and the bill provides guidelines for such procedures. The bill permits the department to establish an archival specimen bank for public health purposes, and to establish criteria for the evaluation of requests to use archived specimens for research consistent with public health purposes. Research use of specimens must comply with federal research regulations. The department may require internal review board approval before approving research requests and may charge reasonable fees for the evaluation of requests and the use of archived specimens.

Explanation of amendments, if any: The committee amendment (AM 91) rewrites subsection (4) of section 71-519 as amended in the bill. The committee amendment grants authority to the Department of Health and Human Services Regulation and Licensure over the use, retention, and disposal of blood specimens collected in connection with metabolic disease testing.

The department is required to adopt and promulgate rules and regulations relating to the retention and disposal of such specimens. The rules and regulations must consistent with nationally recognized standards for laboratory accreditation and must comply with all applicable provisions of federal law. The disposal must be conducted in the presence of a witness, and a written or electronic record of the disposal, verified by the witness, must be maintained.

The department must also adopt and promulgate rules and regulations relating to the use of such specimens. Use may only be made for public health purposes with the written consent of the parent or guardian of the infant from whom the specimen was derived, and any such use must comply with all applicable provisions of federal law. The department is permitted to charge a reasonable fee for evaluating research proposals for the use of the specimens and for preparing and supplying specimens for research uses approved by the department.

The committee amendment clarifies the authority of the department with respect to blood specimens obtained in connection with metabolic disease testing and requires the establishment of standards for the retention, use, and disposal of such specimens in rule and regulation.

APPENDIX K

Nebraska Statutes § 44-2816, 44-2822, 71-519 to 71-524

44-2816
Informed consent, defined.

Informed consent shall mean consent to a procedure based on information which would ordinarily be provided to the patient under like circumstances by health care providers engaged in a similar practice in the locality or in similar localities. Failure to obtain informed consent shall include failure to obtain any express or implied consent for any operation, treatment, or procedure in a case in which a reasonably prudent health care provider in the community or similar communities would have obtained an express or implied consent for such operation, treatment, or procedure under similar circumstances.

Source:
Laws 1976, LB 434, § 16.

Annotations:
The standard of care in a medical malpractice or negligence action based on inadequate information for a patient's consent to an operation, treatment, or procedure is not determined by a defendant physician's personal or customary routine, but, rather, is based on information which physicians ordinarily supply to patients in like circumstances in the locality or similar localities. Under this section, Nebraska has adopted a "professional" theory, under which expert evidence is indispensable to establish what information would ordinarily be provided under the prevailing circumstances by physicians in the relevant and similar localities. Eccleston v. Chait, 241 Neb. 961, 492 N.W.2d 860 (1992).

The language of this statute is adopted for the purposes of malpractice actions against chiropractors. Jones v. Malloy, 226 Neb. 559, 412 N.W.2d 837 (1987).

Because of the definition of "informed consent" as outlined in this provision, the Legislature has committed this state to the "professional" theory of the duty of a physician to disclose the risks of a treatment. The professional theory provides that expert evidence is necessary to determine if the physician acted the same as a reasonable medical practitioner under the same or similar circumstances and similar locality. Smith v. Weaver, 225 Neb. 569, 407 N.W.2d 174 (1987).

44-2822
Claim for bodily injury or death; petition or complaint; file; damages.

Subject to the requirements of sections 44-2840 to 44-2846, a patient or his or her representative having a claim under the Nebraska Hospital-Medical Liability Act for bodily injury or death on account of alleged malpractice, professional negligence, failure to provide care, breach of contract, or other claim based upon failure to obtain informed consent for an operation or treatment may file a petition or complaint in any court of law having requisite jurisdiction. No dollar amount or figure shall be included in the demand in any malpractice petition or complaint, but the petition shall ask for such damages as are reasonable in the premises.

Source:
Laws 1976, LB 434, § 22; Laws 1984, LB 692, § 6.

71-519
Screening test; duties; disease management; duties; immunity from liability.

(1) All infants born in the State of Nebraska shall be screened for phenylketonuria, primary hypothyroidism, biotinidase deficiency, galactosemia, hemoglobinopathies, medium-chain acyl co-a dehydrogenase (MCAD) deficiency, and such other metabolic diseases as the Department of Health and Human Services may from time to time specify. Confirmatory tests shall be performed if a presumptive positive result on the screening test is obtained.

(2) The attending physician shall collect or cause to be collected the prescribed blood specimen or specimens and shall submit or cause to be submitted the same to the laboratory designated by the department for the performance of such tests within the period and in the manner prescribed by the department. If a birth is not attended by a physician and the infant does not have a physician, the person registering the birth shall cause such tests to be performed within the period and in the manner prescribed by the department. The laboratory shall within the period and in the manner prescribed by the department perform such tests as are prescribed by the department on the specimen or specimens submitted and report the results of these tests to the physician, if any, the hospital or other birthing facility or other submitter, and the department. The laboratory shall report to the department the results of such tests that are presumptive positive or confirmed positive within the period and in the manner prescribed by the department.

(3) The hospital or other birthing facility shall record the collection of specimens for tests for metabolic diseases and the report of the results of such tests or the absence of such report. For purposes of tracking, monitoring, and referral, the hospital or other birthing facility shall provide from its records, upon the department's request, information about the infant's and mother's location and contact information, and care and treatment of the infant.

(4) The department shall do all of the following in regard to the blood specimens taken for purposes of conducting The tests required under subsection (1) of this section:

(a) Develop a schedule for the retention and disposal of the blood specimens used for the tests after the tests are completed. The schedule shall meet the following requirements:

(i) Be consistent with nationally recognized standards for laboratory accreditation and federal law;

(ii) Require that the disposal be conducted in the presence of a witness. For purposes of this subdivision, the witness may be an individual involved in the disposal or any other individual; and

(iii) Require that a written record of the disposal be made and kept and that the witness sign the record; and

(b) With the written consent of the parent or legal guardian of the infant, allow the blood specimens to be used for medical research during the retention period as long as the medical research is conducted in a manner that preserves the confidentiality of the test subjects and is consistent to protect human subjects from research risks under subpart A of part 46 of 45 C.F.R., as such regulations existed on September 1, 2001.

(5) The department shall prepare written materials explaining the requirements of this section. The department shall include the following information in the pamphlet:

(a) The nature and purpose of the testing program required under this section, including, but not limited to, a brief description of each condition or disorder listed in subsection (1) of this section;

(b) The purpose and value of the infant's parent, guardian, or person in loco parentis retaining a blood specimen obtained under subsection (6) of this section in a safe place;

(c) The department's schedule for retaining and disposing of blood specimens developed under subdivision (4)(a) of this section; and

(d) That the blood specimens taken for purposes of conducting the tests required under subsection (1) of this section may be used for medical research pursuant to subdivision (4)(b) of this section.

(6) In addition to the requirements of subsection (1) of this section, the attending physician or person registering the birth may offer to draw an additional blood specimen from the infant. If such an offer is made, it shall be made to the infant's parent, guardian, or person in loco parentis at the time the blood specimens are drawn for purposes of subsection (1) of this section. If the infant's parent, guardian, or person in loco parentis accepts the offer of an additional blood specimen, the blood specimen shall be preserved in a manner that does not require special storage conditions or techniques, including, but not limited to, lamination. The attending physician or person making the offer shall explain to the parent, guardian, or person in loco parentis at the time the offer is made that the additional blood specimen can be used for future identification purposes and should be kept in a safe place. The attending physician or person making the offer may charge a fee that is not more than the actual cost of obtaining and preserving the additional blood specimen.

(7) The person responsible for causing the tests to be performed under subsection (2) of this section shall inform the parent or legal guardian of the infant of the tests and of the results of the tests and provide, upon any request for further information, at least a copy of the written materials prepared under subsection (5) of this section.

(8) Dietary and therapeutic management of the infant with phenylketonuria, primary hypothyroidism, biotinidase deficiency, galactosemia, hemoglobinopathies, MCAD deficiency, or such other metabolic diseases as the department may from time to time specify shall be the responsibility of the child's parent, guardian, or custodian with the aid of a physician selected by such person.

(9) Except for acts of gross negligence or willful or wanton conduct, any physician, hospital or other birthing facility, laboratory, or other submitter making reports or notifications under sections 71-519 to 71-524 shall be immune from criminal or civil liability of any kind or character based on any statements contained in such reports or notifications.

Source:
Laws 1987, LB 385, § 1; Laws 1988, LB 1100, § 99; Laws 1996, LB 1044, § 502; Laws 1998, LB 1073, § 85; Laws 2001, LB 432, § 10; Laws 2002, LB 235, § 1.Effective date July 20, 2002.

71-520
Food supplement and treatment services program; authorized; fees.

The Department of Health and Human Services shall establish a program to provide food supplements and treatment services to individuals suffering from the metabolic diseases set forth in section 71-519. To defray or help defray the costs of any program which may be established by the department under this section, the department may prescribe and assess a scale of fees for the food supplements. The maximum prescribed fee for food supplements shall be no more than the actual cost of providing such supplements. No fees may be charged for formula, and up to two thousand dollars of pharmaceutically manufactured food supplements shall be available to an individual without fees each year.

Source:
Laws 1987, LB 385, § 2; Laws 1996, LB 1044, § 503; Laws 1997, LB 610, § 1; Laws 1998, LB 1073, § 86; Laws 2002, LB 235, § 2. Effective date July 20, 2002.

71-521
Tests and reports; department; duties.

The Department of Health and Human Services shall prescribe the tests, the test methods and techniques, and such reports and reporting procedures as are necessary to implement sections 71-519 to 71-524.

Source:
Laws 1987, LB 385, § 3; Laws 1996, LB 1044, § 504; Laws 2002, LB 235, § 3.
Effective date July 20, 2002.

71-522
Central data registry; department; duties; use of data.

The Department of Health and Human Services shall establish and maintain a central data registry for the collection and storage of reported data concerning metabolic diseases. The department shall use reported data to ensure that all infants born in the State of Nebraska are tested for diseases set forth in section 71-519 or by rule and regulation. The department shall also use reported data to evaluate the quality of the statewide system of newborn screening and develop procedures for quality assurance. Reported data in anonymous or statistical form may be made available by the department for purposes of research.

Source:
Laws 1987, LB 385, § 4; Laws 1996, LB 1044, § 505; Laws 1998, LB 1073, § 87; Laws 2002, LB 235, § 4.
Effective date July 20, 2002.

71-523
Departments; powers and duties; adopt rules and regulations; contracting laboratories; requirements; fees.

(1) The Department of Health and Human Services shall provide educational and resource services regarding metabolic diseases to persons affected by sections 71-519 to 71-524 and to the public generally.

(2) The Department of Health and Human Services, the Department of Health and Human Services Finance and Support, and the Department of Health and Human Services Regulation and Licensure may apply for, receive, and administer assessed fees and federal or other funds which are available for the purpose of implementing sections 71-519 to 71-524 and may contract for or provide services as may be necessary to implement such sections.

(3) The Department of Health and Human Services shall adopt and promulgate rules and regulations to implement sections 71-519 to 71-524.

(4) The Department of Health and Human Services shall contract, following competitive bidding, with a single laboratory to perform tests, report results, set forth the fee the laboratory will charge for testing, and collect and submit fees pursuant to sections 71-519 to 71-524. The department shall require the contracting laboratory to: (a) Perform testing for all of the diseases pursuant to section 71-519 and in accordance with rules and regulations adopted and promulgated pursuant to this section, (b) maintain certification under the federal Clinical Laboratories Improvement Act of 1967, 42 U.S.C. 263a, as such act and section existed on July 20, 2002, (c) participate in appropriate quality assurance proficiency testing programs offered by the Centers for Disease Control and Prevention of the United States Department of Health and Human Services or other professional laboratory organization, as determined by the Department of Health and Human Services, (d) maintain sufficient contingency arrangements to ensure testing delays of no longer than twenty-four hours in the event of natural disaster or laboratory equipment failure, and (e) charge to the hospital, other birthing facility, or other submitter the fee provided in the contract for laboratory testing costs and the administration fee specified in subsection (5) of this section. The administration fee collected pursuant to such subsection shall be remitted to the Department of Health and Human Services Finance and Support.

(5) The Department of Health and Human Services shall set an administration fee of not more than ten dollars. The department may use the administration fee to pay for the costs of the central data registry, tracking, monitoring, referral, quality assurance, program operation, program development, program evaluation, and treatment services authorized under sections 71-519 to 71-523. The fee shall be collected by the contracting laboratory as provided in subdivision (4)(e) of this section.

(6) Fees collected for the department pursuant to sections 71-519 to 71-523 shall be remitted to the State Treasurer for credit to the Department of Health and Human Services Finance and Support Cash Fund.

Source:
Laws 1987, LB 385, § 5; Laws 1996, LB 1044, § 506; Laws 1997, LB 610, § 2; Laws 1998, LB 1073, § 88; Laws 2002, LB 235, § 5.
Effective date July 20, 2002.

71-524
Enforcement; procedure.

In addition to any other remedies which may be available by law, a civil proceeding to enforce section 71-519 may be brought in the district court of the county where the infant is domiciled or found. The attending physician, the hospital or other birthing facility, the Attorney General, or the county attorney of the county where the infant is domiciled or found may institute such proceedings as are necessary to enforce such section. It shall be the duty of the Attorney General or the county attorney to whom the Director of Regulation and Licensure reports a violation to cause appropriate proceedings to be initiated without delay. A hearing on any action brought pursuant to this section shall be held within seventy-two hours of the filing of such action, and a decision shall be rendered by the court within twenty-four hours of the close of the hearing.

Source:
Laws 1987, LB 385, § 6; Laws 1996, LB 1044, § 507; Laws 2002, LB 235, § 6.
Effective date July 20, 2002.

APPENDIX L

October 2002
National Newborn Screening and Genetics Resource Center
Site Visit Report

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